



MedGen: A Medical Genetics Portal

A public web portal providing access to medical genetics information available from NCBI
<https://www.ncbi.nlm.nih.gov/medgen>

National Center for Biotechnology Information • National Library of Medicine • National Institutes of Health • Department of Health and Human Services

Scope and Access

MedGen provides up to date information about human disorders and other phenotypes with a genetic component. This database integrates information from multiple sources, such as OMIM, SNOMED CT, and Human Phenotype Ontology, and thus conveys community standards for terms, definitions, descriptions, and identifiers. The content is augmented by the inclusion of professional guidelines curated by staff of the Genetic Testing Registry (GTR), as well as links to publications related to disorders, clinical studies and systematic reviews based on PubMed's Clinical Queries. Data in MedGen are acquired both programmatically and via curation, depending on the type of information and the sources for that information. The first step in organizing the information is to establish a concept that defines a disorder or phenotype, classify that concept by type, and then assign that concept a stable unique identifier. With that framework established, data are connected to that concept.

MedGen is integrated with other biological, clinical and genetic testing resources at NCBI, e.g. the Gene database, the Genetic Testing Registry (GTR), ClinVar, GeneReviews, and Medical Genetics Summaries on NCBI's bookshelf. Data from MedGen can be searched on the web through its homepage (<https://www.ncbi.nlm.nih.gov/medgen>) or accessed programmatically via E-utilities. MedGen provides links to many resources to facilitate discovery. Terms and their relationships are available for download from MedGen's ftp site (<ftp://ftp.ncbi.nlm.nih.gov/pub/medgen/>).

Searching in MedGen

Searching for information in MedGen can be done by typing desired query terms in the search box followed by clicking the "Search" button (A). For more specific data retrieval, field-limited terms connected with appropriate Boolean operators (AND, OR, NOT) are strongly recommended. The "Example searches" section (B) provides a few representative examples of field-limited terms. Clicking an underlined example term retrieves a set of current records. Links to documents on MedGen and relevant tools/resources from NCBI are listed in the middle section (C). The "Limits" (D) links to the Limits page, which provides access to a set of pre-set fields to allow quick retrieval of records with certain characteristics. Advanced" (E) links to a query construction page, where sophisticated queries can be constructed using Boolean operator connected terms limited to selected field for more specific data interrogation.

Search results are displayed in the summary format (F). The information contained within a record can be gauged by the data source (G) and the number of connections to relevant records in other resources (H). Clicking the hyperlinked title of a record opens it in full report format.

The screenshot displays the MedGen homepage with various navigation and search elements. At the top, there's a search bar with a dropdown menu set to 'MedGen' and a 'Search' button. Below the search bar, there are links for 'Limits', 'Advanced', and 'Help'. The main content area is divided into three columns: 'Using MedGen' (with links like 'MedGen Quick Start', 'List of Professional Guidelines', 'Help', etc.), 'MedGen Tools' (with '1000 Genomes Browser' and 'Variation'), and 'Other Resources' (with 'ClinVar', 'Gene', 'Genetic Testing Registry (GTR®)', 'GeneReviews®', 'OMIM®', and 'RefSeqGene'). A red bracket groups the 'Other Resources' section. Below these columns is the 'Example searches' section, which includes a table with columns for 'Name', 'Related gene', and 'Clinical feature'. The first row shows 'achondroplasia[title]' as an example. Below this, there's a search bar with 'Huntington chorea' entered and a 'Search' button. The search results are displayed in a summary format, showing a list of items with their titles and brief descriptions. The first item is 'Huntington chorea' (HD), described as a progressive disorder of motor, cognitive, and psychiatric disturbances. The second item is 'FH: Huntington chorea'. The third item is 'Dementia due to Huntington chorea'. Each item has a link to its full report and a list of associated resources (GTR, ClinVar, Genes, OMIM, GeneReviews).

NCBI Resources How To Sign in to NCBI

MedGen MedGen Search

Limits Advanced Help

MedGen
Organizes information related to human medical genetics, such as attributes of conditions with a genetic contribution.

Using MedGen
MedGen Quick Start
List of Professional Guidelines
Help
MedGen Chapter in The NCBI Handbook
Select condition and phenotype terms for ClinVar and GTR
Frequently asked questions
Downloads/FTP
MedGen News

MedGen Tools
1000 Genomes Browser
Variation

Other Resources
ClinVar
Gene
Genetic Testing Registry (GTR®)
GeneReviews®
OMIM®
RefSeqGene

Example searches

Name	Related gene	Clinical feature
achondroplasia[title]	LMNB1[gene]	sh

As you type your query, names of genetic disorders used in the NIH Genetic Testing Registry (GTR) will be provided. If you do not make a selection from the menu that appears under the search box as you type, your query is processed by looking for a match on a word or phrase. * is used as the wild card, and that wild card can be used only at the end of a word.

If you enter a gene symbol followed by [gene], the diseases caused by or with some association to that gene will be retrieved.

MedGen MedGen Huntington chorea Search

Create alert Limits Advanced

Summary Search results Items: 5

- ☐ **Huntington chorea**
1. Huntington disease (HD) is a progressive disorder of motor, cognitive, and psychiatric disturbances. The mean age of onset is 35 to 44 years and the median survival time is 15 to 18 years after onset. [from GeneReviews]
MedGen UID: 5654 • Concept ID: C0020179 • Disease or Syndrome
GTR ClinVar Genes OMIM GeneReviews
- ☐ **FH: Huntington chorea**
2. MedGen UID: 635299 • Concept ID: C0481629 • Finding
GTR ClinVar Genes OMIM GeneReviews
- ☐ **Dementia due to Huntington chorea**
3. MedGen UID: 536662 • Concept ID: C0236961 • Mental or Behavioral Dysfunction
GTR ClinVar Genes OMIM GeneReviews

Information Provided by a MedGen Record

Clicking the title of a search retrieved record opens it in the “full report” display format (A). This report is organized into clearly marked sections, each addressing a specific aspect of the disease. The “Table of contents” (B) provides a quick way to navigate to sections of interest. Contents under these sections will vary for individual records. MedGen records are integrated with other public resources and available information is organized as links under the “Table of contents.” Here genetic tests for the disease are collected in the “Genetic Testing Registry” section (C). Related records from other

NCBI resources are in the “Related information” section at the end of the right-hand column (not shown). For Huntington's Chorea:

- ◆ The disease synonyms, SNOMED IDs, and gene information are given at the top (D).
- ◆ Phenotypic description of the disease are presented in the “Disease characteristics,” “Additional descriptions,” and “Clinical features” section (E).
- ◆ Two sets of hierarchical terms for the disease are listed under the “Term hierarchy” section (F).
- ◆ Testing and treatment guidelines and updated information from recent clinical studies retrieved using the PubMed Clinical Queries system are displayed in their own sections (G, collapsed).

Display Settings: ☒ Full Report **A**

Huntington's chorea (HD)
MedGen UID: 5654 • Concept ID: C0020179 • Disease or Syndrome

Synonyms: HD; Huntington chorea; Huntington Chronic Progressive Hereditary Chorea; Huntington Disease; Huntington's disease; Progressive Chorea, Chronic Hereditary (Huntington)

Modes of inheritance: Autosomal dominant inheritance

SNOMED CT: HC - Huntington chorea (58756001); HD - Huntington chorea (58756001); Huntington chorea (58756001); Huntington's chorea (58756001); Chronic progressive chorea (58756001); Chronic progressive hereditary chorea (58756001)

Gene: HTT

Cytogenetic location: 4p16.3

OMIM: 143100

Disease characteristics **D**

Excerpted from the GeneReview: Huntington Disease
Huntington disease (HD) is a progressive disorder of motor, cognitive, and psychiatric disturbances. The mean age of onset is 35 to 44 years and the median survival time is 15 to 18 years after onset. [from GeneReviews]

Full text of GeneReview (by section):
Summary | Diagnosis | Clinical Description | Differential Diagnosis | Management | Genetic Counseling | Resources | Molecular Genetics | References | Chapter Notes

Authors: Simon C Warby | Rona K Graham | Michael Hayden view full author information

Additional descriptions **E**

From OMIM
Huntington disease (HD) is an autosomal dominant progressive neurodegenerative disorder with a distinct phenotype characterized by chorea, dystonia, incoordination, cognitive decline, and behavioral difficulties. There is progressive, selective neural cell loss and atrophy in the caudate and putamen. Walker (2007) provided a detailed review of Huntington disease, including clinical features, population genetics, molecular biology, and animal models. <http://www.omim.org/entry/143100>

From GHR
Huntington disease is a progressive disorder characterized by motor, cognitive, and psychiatric problems, and loss of thinking and memory. The disease is usually fatal.

Table of contents **B**

Disease characteristics
Additional descriptions
Clinical features
Term Hierarchy
Professional guidelines
Recent clinical studies
Recent systematic reviews

Genetic Testing Registry **C**

Linkage analysis (8)
Microsatellite instability testing (MSI) (2)
Sequence analysis of the entire coding region (2)
Targeted mutation analysis (13)
Targeted variant analysis (27)
See all (84)

Molecular resources

OMIM
RefSeqGene

Consumer resources

Genetics Home Reference
Genetic Alliance
NCATS Office of Rare Diseases Research (GARD)

<https://www.ncbi.nlm.nih.gov/medgen/5654>

Clinical features

Show all Hide all

● Abnormality of the eye

● Abnormality of the nervous system

- Bradykinesia
- Chorea
- Dementia
- Gliosis
- Personality change
- Rigidity

Term Hierarchy

GTR MedGen

- Disorder by Site
 - Disorder of nervous system
 - Disorder of the central nervous system
 - Movement disorder
 - dyskinesia
 - Chorea
 - Huntington's chorea
 - Juvenile onset Huntington's disease
 - Westphal disease

Professional guidelines

Recent clinical studies

Recent systematic reviews

Chorea **I**

MedGen UID: 505081 • Concept ID: CN001874 • Finding

Chorea (Greek for 'dance') refers to widespread, arrhythmic involuntary movements of a forcible, jerky and restless fashion.

See: Feature record | [Search on this feature](#) **J**

Spinocerebellar ataxia 1

2. Spinocerebellar ataxia type 1 (SCA1) is characterized by progressive cerebellar ataxia, dysarthria, and eventual deterioration of bulbar functions. Early in the disease, affected individuals may have gait disturbance, slurred speech, difficulty with balance, brisk deep tendon reflexes, hypermetric saccades, nystagmus, and mild dysphagia. Later signs include slowing of saccadic velocity, development of up-gaze palsy, dysmetria, dysidiadochokinesia, and hypotonia. In advanced stages, muscle atrophy, decreased deep tendon reflexes, loss of proprioception, cognitive impairment (e.g., frontal executive dysfunction, impaired verbal memory), chorea, dystonia, and bulbar dysfunction are seen. Onset is typically in the third or fourth decade, although childhood onset and late adult onset have been reported. Those with onset over age 60 years may manifest a pure cerebellar phenotype. Interval from onset to death varies from ten to 30 years; individuals with juvenile onset show more rapid progression and more severe disease. Anticipation is observed. An axonal sensory neuropathy detected by electrophysiologic testing is common; brain imaging typically shows cerebellar and brain stem atrophy. [from GeneReviews]

MedGen UID: 155703 • Concept ID: C0752120 • Disease or Syndrome

[GTR](#) [Genes](#) [OMIM](#) [GeneReviews](#)

Spinocerebellar ataxia 7

Entries under “Clinical feature” allow searching by specific clinical features using controlled vocabularies. For example, clicking the arrow in front of “Abnormality of the nervous system” (H) reveals the controlled term Chorea. Clicking the term (I) opens a popup with more details on the term (from HPO). Clicking “Search on this feature” (J) retrieves other conditions (MedGen records) with this clinical feature, highlighting the fact that a given clinical feature is present in different conditions.

Help Documents and MedGen Specific Help

MedGen online help documentation and handbook contain additional details:

<http://www.ncbi.nlm.nih.gov/medgen/docs/help/> and <http://www.ncbi.nlm.nih.gov/books/NBK159970/>

Questions and comments should be addressed to: medgen_help@nih.gov